TITLE: A STUDY ON THE *GJB2, GJB3* AND *GJB6* GENE MUTATIONS AMONG MALAYS WITH NON-SYNDROMIC HEARING LOSS

RESEARCH CENTRE: Human Genome Centre
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CURRENT STATUS OF PROJECT: Ongoing

RESEARCHERS:
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TRACK RECORD:

1. Hearing screening of infants in Neonatal Unit, Hospital Universiti Sains Malaysia using transient evoked otoacoustic emissions (2005)
2. Auditory Neuropathy in a Non High Risk for Hearing Impairment Child: A case report
3. An Evaluative Study of Distraction Test and TEOAE in Detection of Hearing Impairment in Early Childhood

Introduction:

Hearing loss (Hl) can be caused by environmental factors as well as genetic factors. Genetic causes representing 50-70% of Hl with autosomal recessive inheritance which representing approximately 80% of this total. Many genes are involved and several of these genes have been identified and three of them are GJB2, GJB3 and GJB6 gene.

Objectives:

1. To identify the GJB2, GJB3 and GJB6 gene mutations in Malay patients with non-syndromic hearing loss and normal hearing
2. To determine the association between the GJB2, GJB3 and GJB6 gene mutations and non-syndromic hearing loss
3. To determine the association between the GJB2, GJB3 and GJB6 gene mutations with the clinical phenotype

Methodology:

Genomic DNA will be extracted from buccal swab. The extracted DNA will undergo Polymerase Chain Reaction (PCR) to amplify the GJB2, GJB3 and GJB6 coding region. The PCR product will be sequenced to determine the mutations. The known and unknown mutations will be detected by denaturing High Performance Liquid Chromatography (dHPLC). For statistical analysis, SPSS version 11.0 will be used for data entry and data analysis.

Expected Outcome:

The mutations in GJB2, GJB3 and GJB6 gene in Malay patients with non-syndromic hearing loss can be identified and the association between those genes can be determined. Also the association between the genes mutation with the clinical phenotype will be determined.